

The National Registry for Ichthyosis and Related Disorders Will Cease Operation after 2012

The National Registry for Ichthyosis and Related Disorders (the Registry) was funded in 1994 by the National Institute for Arthritis and Musculoskeletal and Skin Diseases of the National Institutes of Health (NIH) to improve understanding of the diagnosis, pathophysiology, and treatment of the ichthyoses. The request for applications was the result of lobbying by members of the Foundation for Ichthyosis and Related Skin Types (FIRST; <http://www.firstskinfoundation.org>), a lay group that supports individuals with ichthyosis and their families. The Registry application was the product of collaboration among members of the Medical and Scientific Advisory Board (MSAB) of FIRST, and the MSAB has continued to provide advice since inception of the Registry. Over 11 years of funding, 611 affected individuals were enrolled and characterized clinically. Review of dermatopathology specimens, measurement of serum cholesterol sulfate levels, and mutation detection were offered when appropriate. Each subject's response to a quality-of-life questionnaire was determined at the time of enrollment. Information was entered into a secure database by the registered nurse who interviewed enrollees and reviewed by the principal investigator for accuracy and clarification. Enrollees were contacted annually to inform them of the Registry's progress and to update contact information.

NIH funding ceased in 2004. At that time enrollment stopped and the Registry continued on a maintenance basis, providing access to information obtained during enrollment. The Registry has served its purpose well. Support for investigators has included informing appropriate enrollees of studies, data mining, correlations of histology with phenotype, genotype-phenotype correlation, and searching for new mutations. Twenty studies (Figure 1) resulting in 22 publications have been/are being

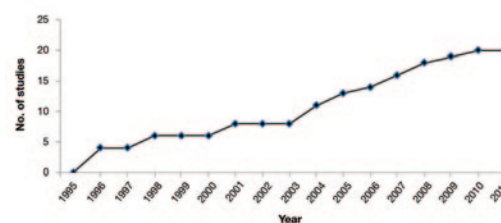


Figure 1. Number of studies, by year, supported by the National Registry for Ichthyosis and Related Disorders.

supported (complete information on studies supported and ensuing publications is available at <http://www.skinregistry.org>). Two of these studies are active, and data for a new project are being reviewed by the principal investigator.

FIRST, which has supported the Registry throughout its existence, has determined that research is now being conducted effectively in other ways. The wide acceptance and use of the Internet and social networking, as well as advances in other technologies and methodologies, have now altered the value of the original Registry concept. Thus, although maintenance of the Registry is relatively inexpensive, it cannot grow in its current form. FIRST will seek and support new initiatives associated with their research programs, and funding for the Registry will cease at the end of 2012.

Investigators who wish to utilize the Registry should contact the principal investigator via the website. Support for investigators wishing to utilize the information in the Registry database will be limited after 2012. We encourage investigators to contact the Registry via the website in order to take advantage of this unique resource while it is available.

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